

# Molecular Diagnostics

## 1. Immunoglobulin Heavy Chain (IgH) Gene Rearrangement by PCR

Early in B-cell development, the DNA of the immunoglobulin heavy chain (IgH) locus undergoes a series of rearrangements. These create unique combinations of IgH DNA that can be used as a clonal marker of the cell progeny. The test is useful in the diagnosis or confirmation of B-cell clonal disorders.

### About the test

The assay utilizes the Polymerase Chain Reaction (PCR) in conjunction with primers that recognize the conserved sequence regions of the IgH framework III (Fr III) variable and joining gene segments to amplify rearranged IgH DNA sequences. Amplified PCR products are resolved by polyacrylamide gel electrophoresis and viewed under ultraviolet light following staining of the DNA with a fluorescent dye.

### Specimen Requirements

EDTA- or ACD-anticoagulated whole blood (min. 2 ml) or bone marrow (min. 1 ml), fresh tissue, frozen tissue or formalin-fixed, paraffin-embedded tissue can be used for this test. Call for specific specimen requirements, handling and shipping information. **Do not separate blood or bone marrow products.**

Order Code	BGENE
CPT Codes	83891, 83894, 83898 (x3) and 83912
Routine TAT	Performed once per week, Mon – Fri. Reported next day.

## 2. T-cell Receptor (TCR) Gene Rearrangement by PCR

During T-cell differentiation, TCR genes undergo a series of rearrangements. These create unique combinations of TCR DNA that can be used as a clonal marker of the cell progeny. This assay amplifies TCR $\gamma$  and TCR  $\beta$  DNA sequences and is useful for diagnosis or confirmation of T-cell clonal disorders.

### About the test

The Polymerase Chain Reaction (PCR) together with primer sets that recognize conserved sequence regions of the variable, diversity and joining gene segments of the TCR $\gamma$  and TCR  $\beta$  genes are utilized to amplify rearranged TCR $\gamma$  and TCR  $\beta$  DNA sequences. Amplified PCR products are resolved by gel electrophoresis and viewed under ultraviolet light following staining of the DNA with a fluorescent dye.

### Specimen Requirements

EDTA- or ACD-anticoagulated whole blood (min. 2 ml) or bone marrow (min. 1 ml), fresh tissue, frozen tissue or formalin-fixed, paraffin-embedded tissue can be used for this test. Call for specific specimen requirements, handling and shipping information. **Do not separate blood or bone marrow products.**

Order Code	TGENE
CPT Codes	83891, 83894, 83898 (x9) and 83912
Routine TAT	Performed once per week, Mon – Fri. Reported next day

## 3. IgH and TCR Gene Rearrangement by PCR

These are the same tests that are described above, but at a discounted price for ordering both. This test can only be ordered on the same specimen.

### Specimen Requirements

EDTA- or ACD-anticoagulated whole blood (min. 2 ml) or bone marrow (min. 1 ml), fresh tissue, frozen tissue or formalin-fixed, paraffin-embedded tissue can be used for this test. Call for specific specimen requirements, handling and shipping information. **Do not separate blood or bone marrow products.**

Order code	BTGENE
CPT Codes	83891, 83894 (x2), 83898 (x11) and 83912
Routine TAT	Performed once per week, Mon – Fri. Reported next day.

**Note:** The Polymerase Chain Reaction (PCR) process is covered by U.S. patents owned by Hoffman-LaRoche Inc. These tests are performed under a licensed agreement with Roche Molecular Systems Inc.

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### 4. HER-2/*neu* Gene Amplification Determination by FISH

The HER-2/*neu* proto-oncogene (also known as *c-erbB-2*, HER2 and *neu*) encodes a 185-kDa transmembrane glycoprotein receptor protein (p185<sup>HER2</sup>) that demonstrates tyrosine kinase activity and has homology to the epidermal growth factor receptor. In human breast cancer, overexpression of the p185<sup>HER2</sup> protein has been reported in approximately 25-35% of lymph node-positive breast cancer cases and is associated with poor prognosis, decreased and/or increased sensitivity to chemotherapy, increased risk of recurrence and shorter overall survival. Thus, HER-2/*neu* gene amplification serves as a powerful prognostic indicator in lymph node-positive breast cancer.

#### About the test

FISH will be performed using the Vysis PathVysion™ HER-2/*neu* Gene Detection System. This system is approved by the United States Food and Drug Administration (FDA) for the determination of HER-2/*neu* gene amplification in formalin-fixed, paraffin-embedded (FFPE) tissue specimens. Samples will be objectively graded by counting the number of HER-2/*neu* gene copies with respect to the number of chromosome 17 control copies.

#### Specimen Requirements

Formalin-fixed, paraffin-embedded tissue block. *Alternatively, tissue sections may be sent. Please contact the Molecular Diagnostics laboratory for further information.* Ship at room temperature.

Order code	HER2
CPT Codes	88271 (x2), 88274 and 88365
Routine TAT	Performed once per week, Mon – Fri. Reported next day.